AUTHOR INDEX VOLUME 19, 1999

Amato, Anthony A., 45

Barcellos, Lisa F., 281 Barohn, Richard J., 45 Berger, Joseph R., 101, 193 Bird, Thomas D., 253 Bouchard, Jean-Pierre, 59 Brais, Bernard, 59 Breakefield, Xandra O., 271 Brewer, George J., 261

Chamberlain, Jeffrey S., 323 Chance, Phillip F., 353 Ciacci, Joseph D., 213 Clifford, David B., 185 Cohen, Bruce A., 201 Cole, N., 407

Di Rocco, Alessandro, 151

Fardeau, M., 59 Fink, John K., 249, 261, 301, 339

Hartigan-O'Connor, Dennis, 323 Hauser, Stephen L., 281 Hayden, Michael R., 385 Hedera, Peter, 261, 301 Hopkins, Linton C., 67 Keller, Marcel P., 353 Kissel, John T., 1, 5, 35 Klein, Christine, 271

Lanska, Douglas J., 105 Leavitt, Blair R., 385 Leppert, Mark F., 397 Levy, Robert M., 213 Lindblad, Kerstin, 289

Major, Eugene O., 193 Marra, Christina M., 177 McArthur, Justin C., 129 McDaniel, D.O., 419 Mendell, Jerry R., 1, 5, 9 Mintz, Mark, 165

Nath, Avi, 113

Oksenberg, Jorge R., 281 Ozelius, Laurie J., 271

Pandolfo, Massimo, 311 Pascuzzi, Robert M., 3, 87, 103, 235, 247 Ptáček, Louis J., 363

Rouleau, Guy A., 59

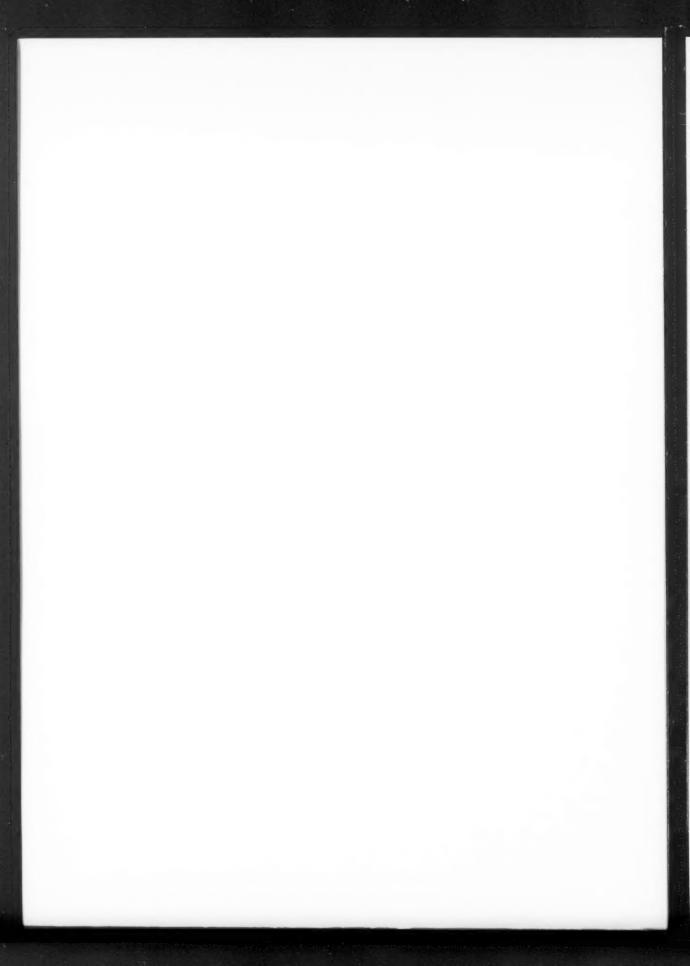
Sacktor, Ned, 129 Schalling, Martin, 289 Selnes, Ola, 129 Shoffner, John M., 341 Siddique, T., 407 Simpson, David M., 157 Singh, Nanda, 397 St. George-Hyslop, P.H., 371 Subramony, S.H., 419

Tawil, Rabi, 81 Tellez, Claudia, 213 Thornton, Charles, 25 Tomé, Fernando M.S., 59 Tsao, Chang-Yong, 9

Vig, P.J.S., 419 VonRoenn, Jamie, 213

Wagener, Maylene E., 67 Warren, Stephen T., 67 Weber, Thomas, 223 Wellington, Cheryl L., 385 Wulff, Enrique A., 157

Zacharias, Alan S., 67



SUBJECT INDEX VOLUME 19, 1999

Acquired immunodeficiency syndrome, 105–110, 113, 129–132, 151–154, 157–161, 165–166, 201–209, 213–219, 223–229

Adenoviral vector technology, 326-327

Adenoviral vectors, transfer to both proliferating and nonproliferating cells using, 84–85

Adjunctive therapies for HIV dementia, 146

Adolescent-onset PTD of mixed type, dystonia 6, 274

Adult-onset focal dystonia, dystonia 7, 275

AIDS, see acquired immunodeficiency syndrome AIDS, incidence, survival and mortality of, 107–108

AIDS, includence, survival and mortality of, 107–108

AIDS-related lymphomas, immunosuppression and the molecular biology of, 213–214

Algorithm for patient diagnosis with OXPHOS defects, 343-345

Alzheimer disease, the mitrochondrial genome and, 379 Amyloid precursor protein, 372–373

Amyotrophic lateral sclerosis, pathology and diagnosis of, genetics of, mechanism of the disease, 411–412

Anabolic steroids, 83

Anticopper drug treatment for Wilson's disease, 265–267 Anticopper drugs, various phases and classes of patients in the application of, 266–267

Antiretroviral therapy, effect on HIV levels, CNS penetration, 142-144

Apolipoprotein E gene, analysis revealed that the gene likely responsible for the genetic locus on chromosome 19 was the, 373–374

Arachidonic acid, metabolites, the synthesis of, 121 Arrhythmia, 69, 73

Asymptomatic at-risk patients, the other major scenario in DNA testing is the genetic evaluation of, 256

Autonomic neuropathy, 162

Autosomal dominant HSP, 303, 305

Autosomal dominant nocturnal frontal lobe epilepsy, 399-400

Autosomal recessive HSP, 302-303, 305

Autosomal recessive PTD, 275

Bacillary angiomatosis, neurologic complications associated with, 180

Becker dystrophy, 5, 12-13, 89

Benign familial neonatal convulsions, 398-399

Biochemistry and genetics of oxidative phosphorylation, 342–343, 345

Blood-brain barrier, mechanism of viral entry across the, 115 Bone marrow cell transplantation, 85

Brain, release of viral particles infecting cells, 115

Candidate genes, in relation to HSP genes, 304 Carriers of EDMD, 73

Caspase inhibition, therapeutic approach for HD, 394

Cellular targets of HIV infection, 139-140

Central nervous system, the effect of Friedreich's ataxia on the, 312

Central nervous system, viral strains in the, entry of HIV into the, 116, 140

Cerebellar ataxia, Leigh disease and, 346-347

Cerebrospinal fluid analysis for the diagnosis of HIV-related neurologic diseases, 223–229

Cerebrospinal fluid, laboratory examinations and investigations of HIV-1 infection in, 171–172 Charcot-Marie-Tooth neuropathy, 354

Chemokine dysregulation in HIV encephalopathy, 116-117

Children, HIV-associated neurologic disease in, 165-174

Children, the DNA testing of, the role of testing in the adoption of, 257–258

Chromosome 12 locus, 378

Chromosome 2p-linked HSP, trinucleotide repeat expansion in, 303-304

Chromosome 4q35, 41

Clinical and genetic evaluation of CMT, 358

Clinical features and host factors associated with PML, 195-196

Clinical presentation of PCNSL, 215

CMT, see Charcot-Marie-Tooth neuropathy, 354

CMT type 1A, 354

CMT type 1B, 354-355

CMV, see cytomegalovirus

CMV encephalitis, 186

CMV neurologic disease, diagnosis of, therapy for, 187–188 CMV radiculomyelitis, it is not rare for encephalitis to be

associated with the presentation of, 186

CNS, see central nervous system CNS lymphoma, 213–214

CNS toxoplasmosis, 204–206

Combination art, effects of the treatment on HIV dementia, 145

Congenital muscular dystrophies, 17-20

Contracture, 68, 73-75

Corticosteroids, 82-83

Costs of DNA-based tests, in relation to other diagnostic tests, 255

Creatine kinase, 11, 17-19, 27, 39

Cryptococcal meningitis, background, clinical manifestations of, diagnosis of, treatment of, 181–182

CTG repeats, 25-28, 30-31

Cytokine dysfunction in HIV encephalopathy, 116

Cytomegalovirus, the most common cause of viral neurologic complications encountered in HIV infection is, 185

Dejerine-Sottas syndrome, 355

Demyelinating neuropathies, other forms of, 357

Depression, a serious risk for persons who have tested positive for a neurogenetic disease is, 256–257

Depression, in children and adolescents with HIV-1 infection, 169

Diabetes mellitus, about 10% of Friedreich's ataxia patients develop, 313-314

Diagnosis and prognosis of DNA testing, 254-255

Diagnosis and treatment of Wilson's disease, 261-268

Diffuse infiltrative lymphocytosis syndrome, clinical presentations of, 162

Distal myopathies, historical perspective, 45-46

Distal symmetric polyneuropathy, clinical and pathologic features as a neurologic complication of AIDS, 158–161

Distal, 45-46, 49-51, 53-55

DMD, see Duchenne muscular dystrophy

DMD and dystrophin, salient features of, 324-325

DNA diagnosis, identification of the CAG expansion mutations and, 422–423

DNA testing, symptomatic patients and, treatment of, 254,

Dominant ataxias, clinical diagnosis of, 420-421

Dominantly inherited ataxias, 419-424

"Domino effect", HIV-infected cells in the brain leading to progressive loss of neuronal function, 122

Duchenne muscular dystrophy, 5-7, 10-15, 87-88, 323-330

Dystonia, the most simple classification subdivides into primary or secondary, 271-272

Dystrophin, 10, 12, 14-16

Dystrophin-glycoprotein complex, core components of, 10 Dystrophinopathies, clinical features, laboratory features, genetics, molecular pathogenesis of the, treatment, 11-15

EBV, see Epstein-Barr virus

EBV as a console factor for PCNSL in association with AIDS, 215

EDMD, see Emery-Dreifuss muscular dystrophy

Electrophysiology, nerve conduction studies in EDMD, 71

Emery-Dreifuss muscular dystrophy, history of, clinical features, cardiac manifestations of, diagnosis of, treatment of, 67-70, 75-76

Endogenous toxins, 121-122

Epidemic, epidemiology of the AIDS virus, 130-131

Epidemiology, of neurologic diseases associated with HIV infection, 109

Episodic disorders affecting excitable tissues, 364 Epstein-Barr virus, 189, 215, 225

Expanded repeat sequences and disease, 289-296

Expanded repeats, methods for detection and isolation of, 295-296

Expanded sequences, repeats and mechanisms of, 290-291

Facioscapulohumeral dystrophy, historical note, clinical features, laboratory features, molecular genetics of, diagnosis of, treatment of, 5-6, 35-42

Familial Alzheimer disease, 378–379

Filamentous intranuclear inclusions, identification of a morphological marker for OPMD

Fragile X mental retardation, the most common form of inherited mental retardation is, 294

Frataxin gene, point mutations in the, 316

Frataxin, the function and pathogenesis of, 316-317

Free radicals, Friedreich's ataxia and the role of, 317

Friedreich's ataxia, clinical aspects and pathogenesis of, 311-318

Friedreich's ataxia, epidemiology of, 312

Friedreich's ataxia, the identification as a triplet repeat disorder, 294

Function disorders in relation to expanded repeats, 293-294

Gene amplification, 223

Gene mutations causing several neurologic disorders, 304 Gene therapy of DMD, requirements for the, 325-326

Gene therapy, Duchenne muscular dystrophy and the progress toward, 323-330

Gene therapy, myoblast transfers for the muscular dystrophies Generalized epilepsy with febrile seizures plus, 400-401 Genetic analysis of HSP, 302-303

Genetic analysis of MS, case-control versus family-based study designs, 283-284

Genetic aspects of multiple sclerosis, 281-287

Genetic disorders of motor neurons, 407-415

Genetic risks, DNA testing has important genetic counseling implications, 255

Genetic susceptibility, its role in the etiology of most MS cases, 281-282

Genetic testing of OXPHOS diseases, 345

Genetics of epilepsy, relevance of findings to clinical

Genetics of primary dystonia, 271-278

Genome searches underscore a role for the MHC, 286-287

Genomic screen, 286-287

Gowers, textbook revisited, early observations on muscular dystrophy, 87-92

Growth factors, growth hormone and insulin-like, 83-84 Gutted adenoviral vectors, 328-329

HD, see Huntington disease

HD patients, recent insights from a human database of, 386-388

Heart disease, in patients with Friedreich's ataxia premature death may be contributed to by, 313

Heart, Friedreich's ataxia and the typical pathology of the, 312-313

Hepatic presentation, recognizing Wilson's disease in patients with, 262-263

Hereditary sensory neuropathies, 357-358

Hereditary spastic paraplegia, 301-307, 410

Herpes simplex virus, another important virus to be considered in the setting of HIV is, 188-189

History of neurology, 235-237

HIV, see human immunodeficiency virus

HIV dementia, viral load and, 115

HIV infection, surveillance of, AIDS as a proxy indicator of,

HIV-associated dementia, clinical and pathologic features of, 131-146

HSP, see hereditary spastic paraplegia

Human herpesvirus-6, 189-190

Human immunodeficiency virus infection, history and prospects, transmission and risk factors of, 105-106

Human immunodeficiency virus type-1, 165-174

Human immunodeficiency virus, 105-110, 113-122 129-146, 151-154, 157-163, 165, 185-190, 202-203, 223-229

Human infection with T. gondii, 202

Huntington disease, clinical and neuropathological features

Idiopathic epilepsies with known gene defects, 398-402 Idiopathic epilepsies with known genetic loci, 402–403

IGF-1, double-blind, placebo-controlled trial in myotonic dystrophy, 84

Imaging characteristics, the diagnosis of PCNSL may be suggested by findings on, 216

In vitro models of HD, recent insights from, 392-394

Infected cells, in relation to HIV, 115-116

Infectious myelopathies, cause of spinal cord disease in HIV infection, 153-154

Inflammatory demyelinating polyneuropathy, a relatively infrequent HIV-associated peripheral neuropathy is.

Inheritance model for multiple sclerosis, 287

Inherited peripheral neuropathy, 353–359

Insurance, the sharing of DNA test results is especially pertinent in the context of, 257

Ion channel diseases in humans, the periodic paralyses definition, 365-367

Ion channel diseases, 363-367

Ischemic myelopathy, noninfectious cause of disease in AIDS, 154

JC virus, the PML disease results from infection with the, 194-195

Kayser-Fleischer rings, an excellent diagnostic tool in neurological presentations are, 263

- Kearns–Sayre and chronic progressive external ophthalmoplegia syndromes, 345–346 Kennedy's disease, 291, 410–411 Kinetics of virologic suppression, 143
- Lactic acidosis, 346
- Laing distal myopathy, 50-51
- Learning disorders, HIV-1-associated CNS disease can frequently lead to, 169
- Leber's hereditary optic neuropathy, 347
- Legal counseling, DNA testing and, 258
- Leigh disease, 348
- Limb-girdle muscular dystrophy, original definition of, clinical features, laboratory features, genetics, molecular pathogenesis of the, treatment, 15–17
- Liver biopsy, the gold standard of Wilson's disease diagnosis is, 264
- Liver disease, the prognosis of Wilson's disease, 268 Lymphoma of the central nervous system in AIDS,
- 213–219

 Major histocompatibility complex, the genetic association
- between MS susceptibility and the, 284–286 Markesbery-Udd distal myopathy, clinical features, laboratory
- features, molecular genetics of, 46–47 *Mdx* mice, prevention of dystrophy in, 326
- Meiotic unequal crossover, genesis of CMT1A and HNPP, 356–357
- Membrane instability, unifying hypothesis of, 11
- Meningitis, 178-179
- Metabolic testing in OXPHOS diseases, 344
- Methods of cerebrospinal fluid analysis, 224
- MHC, see major histocompatibility complex
- Microglial cells, 115
- Migraine headache and epilepsy, the most common episodic phenomena encountered by neurologists are, 364
- Mitochondrial DNA mutations, 345
- Mitochondrial encephalomyopathy, 346
- Miyoshi distal myopathy, clinical features, laboratory features, molecular genetics of, 49–50
- Molecular genetics of Alzheimer disease, 371–379
- Molecular genetics of Friedreich's ataxia, 315-316
- Molecular pathogenesis of Huntington disease, 385–394
- Molecular testing, Friedreich's ataxia and the role of, 314–315
- Monotherapy, effects of the treatment on HIV dementia, 144–145
- Motor neuron degeneration and dementia, 414-415
- MS, see multiple sclerosis
- Multiple sclerosis, gene identification in, 282–283
- Muscle biopsy, in EDMD, 71–73
- Muscle tissue, delivery of adenoviral vectors to, 329-330
- Muscular dystrophy, 5-7, 9-20, 35-42, 45-55, 59-64, 81-85
- Mycobacterial infections, 177-179
- Mycobacterial meningitis, 179
- Mycobacterium tuberculosis, 178-179
- Myoclonic epilepsy and ragged-red fiber disease, 346
- Myofibrillar myopathy, clinical features, laboatory features, molecular genetics of, 51–52
- Myopathies with distal weakness, childhood-onset, muscular dystrophies, inflammatory myopathies, metabolic and congenital myopathies, neuromuscular junction disorders, 52, 55
- Myopathy, associated with HIV, 162-163
- Myotonia, 27-28, 31
- Myotonic dystrophies, genetics of, pathophysiology, clinical features, treatment of, 25–28, 30–31
- Myotonic dystrophy caused by a CTG expansion, 293-294

- Myotonic dystrophy, 5-7, 25-31
- Myotonic myopathy, 25, 30-31
- Negative DNA tests, two cautions must be noted with, 255
- Nervous system, involvement in HIV infection, 108–109
- Neurobehavioral aberrations, increasing evidence in HIV-1 infected children, 168–169
- Neurocognitive deficits in different stages of HIV infection,
- Neuroimaging, radiographic imaging may strongly support the diagnosis of PML, 197
- Neurologic disease, studies of viral load in HIV, 142
- Neurologic diseases related to HIV and AIDS, 109
- Neurologic disturbances due to toxoplasmosis, 205
- Neurologic disturbances due to toxopiasmosis, 205 Neurologic manifestations of toxopiasmosis in AIDS, 201–209
- Neurological manifestations of Wilson's disease, symptomatic treatment of, 267–268
- Neurological presentation, recognizing Wilson's disease in patients with, 263
- Neurological signs and symptoms associated with Friedreich's ataxia, 313
- Neurology trivia, 235–237
- Neuronal cell loss, 116
- Neuropathology and pathogenesis of T. gondii, 202
- Neurophysiological investigations into Friedreich's ataxia, 314
- Neuroradiologic examinations in HIV-1 infection, 171
- Neurosyphilis, 180-181
- Neurotoxicity, tumor necrosis factor-mediated events that contribute to, 121
- Nitric oxide, HIV infection and neurotoxic events, 122
- Nonaka distal myopathy, clinical features, laboratory
- features, relationship to familial inclusion body myopathy, molecular genetics of, 47–49
- Nutritional deficiencies, children with HIV-1 infection are prone to, 170–171
- Oculopharyngeal muscular dystrophy (OPMD), historical background, early descriptions of, clinical definition of, 59–60
- Onset of Friedreich's ataxia, 313
- OPMD, see Oculopharyngeal muscular dystrophy
- Opportunistic infections in relation to HIV-1 infection in older children, 169
- Opportunistic infections, in relation to HIV-related neurological diseases, 224–229
- Oxidative phosphorylation disease diagnosis, 341–349
- OXPHOS, see oxidative disease diagnosis
- OXPHOS disease, nuclear DNA mutations and, 347-348
- PABP2 gene, 59, 62~-64
- Parkinsonism, dystonia associated with, 277
- Paroxysmal dystonia, 275-276
- Pathologic examination of PCNSL, 214
- Pathology and prognosis of PML, 197-198
- PCNSL, see primary CNS lymphomas
- Pelizeaus-Merzbacher disease, proteolipoprotein gene mutations cause, 303-304
- Penicillamine, the first orally effective drug developed for Wilson's disease was, 265–266
- Peripheral neuropathy, most frequent neurologic complication of HIV infection, 158–162
- Pharmacologic therapy, intervention in the muscular dystrophies, 82
- Phenotype-genotype correlations, gene mutations and, 421
- Phenotypes, OXPHOS diseases present with a broad array of,
- 437

SEMINARS IN NEUROLOGY VOLUME 19, NUMBER 4 1999

Plasma HIV levels, 142

PML, see progressive multifocal leukoencephalopathy Polyadenylation binding protein 2 (PABP2) gene, OPMD is

caused by short GCG expansions in the, 60–63 Polyalanine nuclear toxicity, a molecular pathophysiologic hypothesis for OPMD, 63–64

Polyglutamine disorders, the most commonly found pathogenic repeat is CAG, 291–293

Position effect variegation hypothesis, 41

Positional cloning, an alternative strategy to the analysis of individual candidate genes is, 286

Prednisone, 81-83

Prenatal DNA testing, complex issues surrounding, 258

Presenilin 1, 374-376

Presenilin 2, 377-378

Pressure palsies, hereditary neuropathy with liability to, 355 Prevention of HIV infection, 174

Prevention, three levels associated with HIV infection, 109-110

Primary CNS lymphomas, 214-218

Primary dystonia, 274-275

Primary torsion dystonia, early-onset generalized PTD, dystonia 1, 274

Prognosis and treatment for PCNSL, 217

Progressive encephalopathy, associated with neurologic disease in children, 166–168

Progressive multifocal leukoencephalopathy, an overview, 193–194

Progressive myoclonus epilepsy of the Unverricht-Lundborg type, 401-402

Progressive polyradiculopathy, common in HIV-infected patients with advanced immunosuppression, 161–162

PROMM, see proximal myotonic myopathy

Prophylaxis, prevention of toxoplasmic encephalitis recurring having been suppressed, 208–209

Proteolipoprotein, a gene that has been discovered for types of complicated HSP is, 304

Proximal myotonic myopathy, 25, 31

Psychiatric presentation, recognizing Wilson's disease in patients with, 263

Psychiatric symptoms as a reaction to chronic illness, 168–169

PTD, see primary torsion dystonia

Quinolinic acid, levels associated in patients with HIV encephalopathy, 121-122

Radiation therapy in PCNSL, 217-218

Rare monogenic episodic disorders, genetic and biological study of, 364–365

Risks and benefits of DNA testing for neurogenetic disorders, 253-259 Second generation adenoviral vectors, 328

Skeletal muscle pathology in OXPHOS diseases, 344–345 Spinal cord, diseases associated with HIV infection, 151–154

Spinal muscular atrophy, pathology and diagnosis of, genetics of, mechanism of the disease, 407–408

Spinocerebellar ataxia, 291 β-adrenergic agents, 84

Steroid therapy, 82-83

Stress and anxiety related to predictive DNA testing, 256

Susceptibility genes in human epilepsy, 397-403

Systemic aids-related lymphoma with CNS involvement, 219

Terminology describing HIV dementia, 132

Testing for Wilson's disease, 263-264

Therapeutic trials of muscular dystrophy, then . . . and now, 81–82

Therapy in the muscular dystrophies, overview, 81

Therapy of toxoplasmosis in AIDS, 207-208

Tissue biopsy in the diagnosis of PCNSL, 216

Toxoplasma gondii, life cycle of, 201–203, 205–209 Toxoplasmic encephalitis, 203, 206–207

Toxoplasmosis, 201, 205–206

Transgenic mouse model of HD, recent insights from a, 388-391

Treatment of children with HIV-1 infection, 172–174

Trientine, an FDA-approved substitute in patients intolerant of penicillamine is, 266

Trinucleotide, in relation to expanded repeat sequences, 289 Tuberculoma, 178-179

Tumor necrosis factor, implications in the pathogenesis of HIV infection, 121

Urine copper test, diagnosis of Wilson's disease using the, 263

Vacuolar myelopathy, most common spinal cord disease in HIV-infected indiviuals, 151–153

Varicella zoster virus, neurologic manifestations of, 188 Viral latency, seroepidemiologic studies and, 195

Virotoxins, mechanisms of release into extracellular environment, transport of, 117–118

Welander distal myopathy, clinical features, laboratory features, molecular genetics of, 46

Wilson disease, 348

Wilson's disease, recognition of, diagnosis of, 262–263 Wolfram syndrome, 348

X-linked Charcot-Marie-Tooth neuropathy, 357 X-linked HSP, 303, 305-306

Zinc, the most recently FDA-approved drug for the treatment of Wilson's disease is, 265

